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Research Letter

Prenatal diagnosis of a sporadic Apert syndrome by 3-D ultrasound and 3-D helical computerized tomography

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Dear Editor:

We would like to present a fetus with Apert syndrome diagnosed with 3-dimensional (3-D) ultrasonography (USG) and helical computerized tomography.

A 27-year-old woman, gravid 1, was referred to our department at 23 weeks of gestation due to the discovery of clenched fists. Our 2-dimensional (2-D) USG revealed no gap between the parietal and frontal bone on the biparietal diameter planes. Fetal digits of both hands did not move independently. A 3-D USG of the fetal head showed fusion of bilateral coronal sutures, widely open metopic suture and anterior fontanel. "Mitten-hands" were significant on 3-D surface mode reconstruction. These USG findings led to the diagnosis of AS. The parents asked for a 2nd imaging modality. 3-D helical computerized tomography (HCT) was arranged and confirmed bilateral coronal craniosynostosis. All digits were present but in a fixed position compatible with cutaneous syndactyly (Fig. 1). The family chose to terminate the pregnancy and the phenotype of the female abortus was compatible with AS.

Prenatal diagnosis of Apert syndrome (acrocephalosyndactyly type I) is based on the USG findings of abnormal head shape, midfacial hypoplasia, and bony or cutaneous syndactyly of the hands and feet [1]. The skull malformation includes widely separated metopic suture and prematurely ossified coronal sutures. The most challenging part is early diagnosis of craniosynostosis, because coronal synostosis may not be significant and easily overlooked in a routine scan with 2D USG [2]. In our case, craniosynostosis was significant at referral, especially with 3D USG. Though clenched fist was suspected with initial 2D USG, 3D USG provided better recognition of the characteristic "Mitten hand" leading to the diagnosis of AS.

Fetal MRI, without concern of radiation, provides valuable information of associated brain abnormality. However, it is difficult to visualize bony structure anomalies. Contrarily, 3-D HCT can provide additional information of the fetal bones. There is a concern of fetal radiation exposure with 3-D HCT (5.9 mGy), but the mean radiation dose is similar to fetal radiography (3 mGy) [3]. This technique is now widely used in the evaluation of fetal skeletal dysplasia. In our case, 3-D HCT confirmed bilateral coronal craniosynostosis and cutaneous syndactyly.

This disease is inherited in an autosomal dominant manner but most of the cases are sporadic (99.8%). Two missense mutations are recognized to cause Apert syndrome: Ser252Trp and Pro253Arg at the *FGFR2* gene [4]. The high incidence of sporadic cases makes the diagnosis a challenge. Because of pathognomonic imaging features

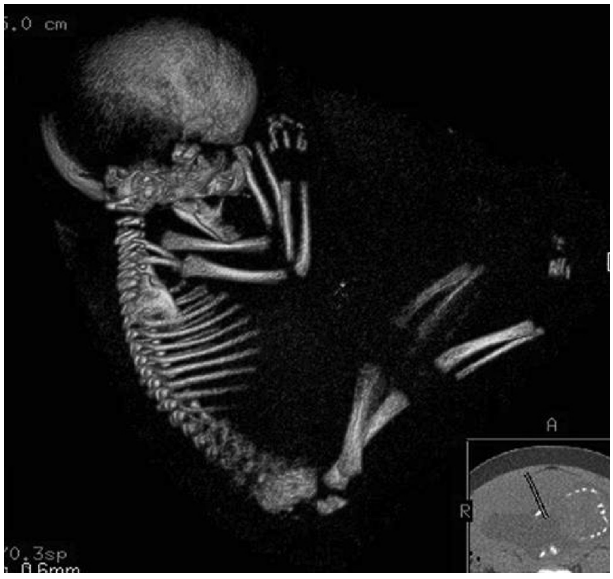

Fig. 1. Fusion of bilateral coronal suture and fixed position of all digits on 3-D helical computerized tomography.

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compatible with AS, the family refused molecular testing or autopsy.

Early diagnosis of sporadic AS relies on early detection of abnormal sonography findings. While the craniosynostosis may be a late presentation, the finding of "Mitten-hands" might raise our suspicion. Prenatal 3-D USG can provide better images than 2-D USG. 3-D HCT can show detailed bony structural information of the whole fetus to confirm the diagnosis and direct subsequent management.

Conflicts of interest

The authors have no conflicts of interest relevant to this article.

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